RUNX1T1 gene

RUNX1 translocation partner 1

Normal Function

The *RUNX1T1* gene provides instructions for making a protein commonly referred to as ETO, which helps regulate the activity of genes. ETO is considered a transcriptional corepressor because it turns off (represses) gene activity. It performs this function by attaching (binding) to proteins that normally turn genes on and blocking their activity. It also interacts with other corepressors to help keep genes turned off.

Health Conditions Related to Genetic Changes

core binding factor acute myeloid leukemia

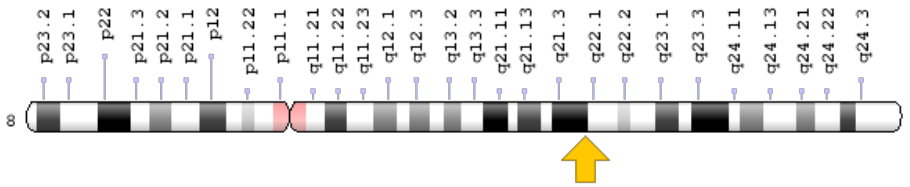
A rearrangement (translocation) of genetic material involving the *RUNX1T1* gene is found in approximately 7 percent of individuals with a form of blood cancer known as acute myeloid leukemia (AML). This translocation, written as t(8;21), combines genetic information from chromosome 8 and chromosome 21, fusing a gene called *RUNX1* on chromosome 21 with the *RUNX1T1* gene on chromosome 8. When associated with this translocation, the condition is classified as core binding factor AML (CBF-AML).

The protein produced from the normal *RUNX1* gene is part of a protein complex known as core binding factor (CBF). As part of CBF, the RUNX1 protein attaches to specific areas of DNA and turns on (activates) genes that are involved in the development of blood cells. Like RUNX1, the fusion protein produced from the t(8;21) translocation, called RUNX1-ETO, attaches to DNA; however, because ETO is involved, the fusion protein turns off genes related to blood cell development instead of turning them on. This change in gene activity blocks the maturation (differentiation) of blood cells and leads to the production of abnormal, immature white blood cells called myeloid blasts. While t(8;21) is important for leukemia development, one or more additional genetic changes are typically needed for the myeloid blasts to develop into cancerous leukemia cells.

Chromosomal Location

Cytogenetic Location: 8q21.3, which is the long (q) arm of chromosome 8 at position 21.3

Molecular Location: base pairs 91,954,967 to 92,103,365 on chromosome 8 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- acute myelogenous leukemia 1 translocation 1, cyclin-D related
- AML1T1
- CBFA2T1
- CDR
- core-binding factor, runt domain, alpha subunit 2; translocated to, 1; cyclin D-related
- eight twenty one protein
- ETO
- MTG8
- MTG8_HUMAN
- myeloid translocation gene on 8q22
- protein CBFA2T1
- runt-related transcription factor 1; translocated to, 1 (cyclin D-related)
- runt related transcription factor 1; translocated to, 1 (cyclin D related)
- zinc finger MYND domain-containing protein 2
- ZMYND2

Additional Information & Resources

Educational Resources

- The Cell: A Molecular Approach (second edition, 2000): Eukaryotic Repressors
  https://www.ncbi.nlm.nih.gov/books/NBK9904/#_A1008_

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28RUNX1T1%5BTI%5D%29+OR+%28ETO%5BTI%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1080+days%22%5Bdp%5D

OMIM

- RUNT-RELATED TRANSCRIPTION FACTOR 1, TRANSLOCATED TO, 1
  http://omim.org/entry/133435

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/ETOID26.html
- HGNC Gene Family: Zinc fingers MYND-type
  http://www.genenames.org/cgi-bin/genefamilies/set/87
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=1535
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/862
- UniProt
  http://www.uniprot.org/uniprot/Q06455

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  http://omim.org/entry/133435

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https://ghr.nlm.nih.gov/gene/RUNX1T1

Reviewed: November 2013
Published: March 21, 2017

Lister Hill National Center for Biomedical Communications
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